

Hyperekplexia Tables 2-5

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 2. Hyperekplexia pathologic allelic variants / *GLRA1*

Sequence change	Mutation	References
del ex1-6	del ex1-6	Brune et al 1996
del ex1-7	del ex1-7	Gilbert et al 2004
G288T / C1030T	W96C / R344X	Tsai et al 2004
G299A	R100H	Coto et al 2005
1 bp delC(601-605) / A830G	1 bp delC(601-605) / M147V	Rees et al 2001
C986A	Y202X	Brune et al 1996, Rees et al 2001
G1030A	R218Q	Miraglia Del Giudice et al 2003
C1073G	S231R	Humeny et al 2002
G1097C	W239C	Gilbert et al 2004
T1112A	I 244N	Rees et al 1994
C1128A	P250T	Saul et al 1999
G1135A / G1555A	R252H / R392H	Vergouwe et al 1999
G1158A	V260M	del Giudice et al 2001
G1178 (T/C)	Q266H	Milani et al 1996
T1188A	S270T	Lapunzina et al 2003
G1192A	R271Q	Elmslie et al 1996, Kwok et al 2001
G1192T	R271L	Rees et al 2001
A1206G	K276E	Elmslie et al 1996, Seri et al 1997
A1216G	Y279C	Kwok et al 2001
1217delT	Y279X	Gilbert et al 2004
A1216C	Y279S	Poon et al 2006
G1406A	G342S	Jungbluth et al 2000, Rees et al 2001

Table 3. Hyperekplexia pathologic allelic variants / *GLRB*

Sequence change	Mutation	Reference
G920A	G229D	
IVS 5+5 G>A	IVS5+5G-A	Rees et al 2002

Table 4. Hyperekplexia pathologic allelic variants / *SLC6A5*

Sequence change	Mutation	Reference
delC{319-324}	P108L fs 25	Rees et al 2006
C916G	L306V	
C1131A	Y377	
C1274T	T425M	
G1294T / Ins[T]1295	V432F/ fs 97	
T1444C	W482R	
A1472G	Y491C	
A1526G	N509S	
C1274T	S510R	
C1888T	Q630X	

Table 5. Hyperekplexia pathologic allelic variants / *GPHN*

Sequence change	Mutation	Reference
A28T	N10Y	Rees et al 2003